

>NM_006920 ACCESSION:NM_006920 NID: gi 29893558 ref NM_006920.2 Homo
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(SCN1A), mRNA
Length = 6046

Score = 4007 bits (10276), Expect = 0.0
Identities = 1996/2009 (99%), Positives = 1996/2009 (99%), Gaps = 0/2009 (0%)
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 Sbjct: 3388 SKEKLNESSSSEGSTVDIGAPVEEQPVVEPEETLEPEACFTEGCVQRFKCCQINVEEGR 3567

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Query: 1741 CDPNKVNPSSSVKGDGCGNPSVGIFFFVSYIIISFLVVVNMYIAVILENFSVATEESAEP 1800
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 Sbjct: 5908 ACPPSYDRVTKPIVEKHEQEGKDEKAKGK 5994



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☐ 1: NM_006920. Homo sapiens sodi...[gi:29893558]

Links

LOCUS NM_006920 6046 bp mRNA linear PRI 05-OCT-2003
 DEFINITION Homo sapiens sodium channel, voltage-gated, type I, alpha (SCN1A), mRNA.
 ACCESSION NM_006920
 VERSION NM_006920.2 GI:29893558
 KEYWORDS .
 SOURCE Homo sapiens (human)
 ORGANISM Homo sapiens
 Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
 Mammalia; Eutheria; Primates; Catarrhini; Hominidae; Homo.
 REFERENCE 1 (bases 1 to 6046)
 AUTHORS Claes,L., Ceulemans,B., Audenaert,D., Smets,K., Lofgren,A.,
 Del-Favero,J., Ala-Mello,S., Basel-Vanagaite,L., Plecko,B.,
 Raskin,S., Thiry,P., Wolf,N.I., Van Broeckhoven,C. and De Jonghe,P.
 TITLE De novo SCN1A mutations are a major cause of severe myoclonic
 epilepsy of infancy
 JOURNAL Hum. Mutat. 21 (6), 615-621 (2003)
 MEDLINE 22637994
 PUBMED 12754708
 REMARK GeneRIF: de novo mutations in SCN1A are a major cause of isolated
 severe myoclonic epilepsy of infancy (SMEI or Dravet syndrome)
 REFERENCE 2 (bases 1 to 6046)
 AUTHORS Sugawara,T., Tsurubuchi,Y., Fujiwara,T., Mazaki-Miyazaki,E.,
 Nagata,K., Montal,M., Inoue,Y. and Yamakawa,K.
 TITLE Nav1.1 channels with mutations of severe myoclonic epilepsy in
 infancy display attenuated currents
 JOURNAL Epilepsy Res. 54 (2-3), 201-207 (2003)
 MEDLINE 22721568
 PUBMED 12837571
 REMARK GeneRIF: SCN1A channels bearing myoclonic epilepsy (SMEI) nonsense
 and missense mutations show attenuated or barely detectable inward
 sodium currents, indicating that SMEI mutations lead to
 loss-of-function and may contribute to development of SMEI
 phenotypes.
 REFERENCE 3 (bases 1 to 6046)
 AUTHORS Chou,I.C., Peng,C.T., Tsai,F.J., Huang,C.C., Shi,Y.R. and Tsai,C.H.
 TITLE The lack of association between febrile convulsions and
 polymorphisms in SCN1A
 JOURNAL Epilepsy Res. 54 (1), 53-57 (2003)
 MEDLINE 22627883
 PUBMED 12742596
 REMARK GeneRIF: the SCN1A gene might not be one of the susceptibility
 factors for febrile convulsions
 REFERENCE 4 (bases 1 to 6046)
 AUTHORS Gennaro,E., Veggiotti,P., Malacarne,M., Madia,F., Cecconi,M.,
 Cardinali,S., Cassetti,A., Cecconi,I., Bertini,E., Bianchi,A.,
 Gobbi,G. and Zara,F.

TITLE Familial severe myoclonic epilepsy of infancy: truncation of Nav1.1 and genetic heterogeneity
JOURNAL Epileptic Disord 5 (1), 21-25 (2003)
MEDLINE 22658066
PUBMED 12773292
REMARK GenerIF: familial severe myoclonic epilepsy of infancy is genetically heterogeneous for SCN1A (Nav1.1) mutation

REFERENCE 5 (bases 1 to 6046)
AUTHORS Weiss, L.A., Escayg, A., Kearney, J.A., Trudeau, M., MacDonald, B.T., Mori, M., Reichert, J., Buxbaum, J.D. and Meisler, M.H.
TITLE Sodium channels SCN1A, SCN2A and SCN3A in familial autism
JOURNAL Mol. Psychiatry 8 (2), 186-194 (2003)
MEDLINE 22497727
PUBMED 12610651
REMARK GenerIF: R542Q in SCN1A was observed in one autism family and had previously been identified in a patient with juvenile myoclonic epilepsy

REFERENCE 6 (bases 1 to 6046)
AUTHORS Ohmori, I., Ouchida, M., Ohtsuka, Y., Oka, E. and Shimizu, K.
TITLE Significant correlation of the SCN1A mutations and severe myoclonic epilepsy in infancy
JOURNAL Biochem. Biophys. Res. Commun. 295 (1), 17-23 (2002)
MEDLINE 22078981
PUBMED 12083760
REMARK GenerIF: Significant correlation of the SCN1A mutations and severe myoclonic epilepsy in infancy

REFERENCE 7 (bases 1 to 6046)
AUTHORS Lossin, C., Wang, D.W., Rhodes, T.H., Vanoye, C.G. and George, A.L. Jr.
TITLE Molecular basis of an inherited epilepsy
JOURNAL Neuron 34 (6), 877-884 (2002)
MEDLINE 22082300
PUBMED 12086636
REMARK GenerIF: The effects of three mutations in SCN1A have been characterized in cultured mammalian cells as a gain-of-function abnormality causing prolonged membrane depolarization, a plausible underlying biophysical mechanism responsible for inherited epilepsy.

REFERENCE 8 (bases 1 to 6046)
AUTHORS Sugawara, T., Mazaki-Miyazaki, E., Fukushima, K., Shimomura, J., Fujiwara, T., Hamano, S., Inoue, Y. and Yamakawa, K.
TITLE Frequent mutations of SCN1A in severe myoclonic epilepsy in infancy
JOURNAL Neurology 58 (7), 1122-1124 (2002)
MEDLINE 21938587
PUBMED 11940708
REMARK GenerIF: Three novel frameshift and seven nonsense mutations in the SCN1A gene have been identified in 14 Japanese patients with severe myoclonic epilepsy in infancy.

REFERENCE 9 (bases 1 to 6046)
AUTHORS Escayg, A., MacDonald, B.T., Meisler, M.H., Baulac, S., Huberfeld, G., An-Gourfinkel, I., Brice, A., LeGuern, E., Moulard, B., Chaigne, D., Buresi, C. and Malafosse, A.
TITLE Mutations of SCN1A, encoding a neuronal sodium channel, in two families with GEFS+2
JOURNAL Nat. Genet. 24 (4), 343-345 (2000)
MEDLINE 20206553
PUBMED 10742094

REFERENCE 10 (bases 1 to 6046)
AUTHORS Malo, M.S., Blanchard, B.J., Andresen, J.M., Srivastava, K., Chen, X.N., Li, X., Jabs, E.W., Korenberg, J.R. and Ingram, V.M.
TITLE Localization of a putative human brain sodium channel gene (SCN1A)

to chromosome band 2q24

JOURNAL Cytogenet. Cell Genet. 67 (3), 178-186 (1994)

MEDLINE [94340991](#)

PUBMED [8062593](#)

REFERENCE 11 (bases 1 to 6046)

AUTHORS Lu, C.M., Han, J., Rado, T.A. and Brown, G.B.

TITLE Differential expression of two sodium channel subtypes in human brain

JOURNAL FEBS Lett. 303 (1), 53-58 (1992)

MEDLINE [92275082](#)

PUBMED [1317301](#)

COMMENT PROVISIONAL REFSEQ: This record has not yet been subject to final NCBI review. The reference sequence was derived from [AY043484.1](#). On Apr 16, 2003 this sequence version replaced gi:[21914835](#).

FEATURES

Location/Qualifiers

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go_component: voltage-gated sodium channel complex [goid 0001518] [evidence IEA];

go_function: cation channel activity [goid 0005261] [evidence IEA];

go_function: voltage-gated sodium channel activity [goid 0005248] [evidence NAS] [pmid 10742094];

go_function: calcium ion binding [goid 0005509] [evidence IEA];

go_process: cation transport [goid 0006812] [evidence IEA];

go_process: sodium ion transport [goid 0006814] [evidence NAS] [pmid 10742094]"

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misc_feature

457..852

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misc_feature

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In some sub-families (e.g. Na channels) the domain is repeated four times, whereas in others (e.g. K channels) the protein forms as a tetramer in the membrane. A bacterial structure of the protein is known for the last two helices but is not the Pfam family due to it lacking the first four helices"

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